SHANK3 gene

SH3 and multiple ankyrin repeat domains 3

Normal Function

The *SHANK3* gene provides instructions for making a protein that is found in many of the body's tissues but is most abundant in the brain. The SHANK3 protein plays a role in the functioning of synapses, which are the connections between nerve cells (neurons) where cell-to-cell communication occurs. Within synapses, the SHANK3 protein acts as a scaffold that connects neurons, ensuring that the signals sent by one neuron are received by another.

The SHANK3 protein is also involved in the formation and maturation of dendritic spines. Dendrites are specialized extensions from neurons that are essential for the transmission of nerve impulses. Dendritic spines are small outgrowths from dendrites that further help transmit nerve impulses and increase communication between neurons.

Health Conditions Related to Genetic Changes

22q13.3 deletion syndrome

The characteristic signs and symptoms of 22q13.3 deletion syndrome, which is also commonly known as Phelan-McDermid syndrome, are caused by a deletion near the end of the long (q) arm of chromosome 22. The chromosomal region that is typically deleted is thought to contain many genes, including the *SHANK3* gene. As a result of the deletion, people with this condition have only one copy of the *SHANK3* gene in each cell instead of the usual two copies.

Researchers believe that a deletion of the *SHANK3* gene and a reduction in the amount of SHANK3 protein produced is responsible for many of the features of 22q13.3 deletion syndrome. A decrease in the functioning of synapses and cell-to-cell communication between neurons caused by a lack of SHANK3 protein is thought to contribute to the developmental delay, intellectual disability, and absent or severely delayed speech characteristic of people with 22q13.3 deletion syndrome.

other disorders

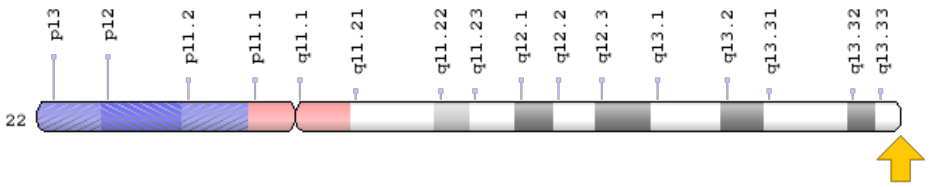
At least seven *SHANK3* gene mutations have been found in people who have the features of autism or similar conditions known as autism spectrum disorders. These disorders are characterized by impaired communication and socialization skills, as well as delayed development of speech and language. Most of these mutations disrupt the function of the SHANK3 protein or lead to the production of an abnormally

short version of the protein. It is unclear how changes in the *SHANK3* gene are related to the risk of developing autism. Researchers suspect that a disruption in nerve cell communication contributes to the development of autism.

Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 50,674,642 to 50,733,212 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- proline-rich synapse-associated protein 2
- ProSAP2
- SPANK-2

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Synaptic Transmission
  https://www.ncbi.nlm.nih.gov/books/NBK11001/

GeneReviews

- Phelan-McDermid Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1198

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SHANK3%5BTIAB%5D%29+OR+%28PROSAP2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

OMIM

- SH3 AND MULTIPLE ANKYRIN REPEAT DOMAINS 3
  http://omim.org/entry/606230

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SHANK3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SHANK3%5Bgene%5D
- HGNC Gene Family: Ankyrin repeat domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/403
- HGNC Gene Family: PDZ domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1220
- HGNC Gene Family: Sterile alpha motif domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/760
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14294
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/85358
- UniProt
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